UK announces mass genome sequencing programme for newborns

December 13 2022

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UK health authorities on Tuesday announced a pioneering research programme using the genomes of 100,000 new-born babies, to detect rare genetic illnesses and speed up treatment.

The £105 million ($129 million) publicly funded "Newborn Genomes Programme"—billed as the largest study of its kind in the world—will establish whether genomic sequencing to diagnose such conditions should be rolled out across the whole country to ensure earlier interventions.

Some 200 conditions affecting a total of 3,000 newborn babies every year in the UK will be screened.

"We will only be looking at conditions that are treatable and early childhood conditions," said Rich Scott, chief medical officer for Genomics England, which was set up by the health ministry in 2013.

They include biotinidase deficiency—a genetic disorder in which the body is unable to metabolise the vitamin biotin.

David Bick, clinical advisor to the programme, said the condition, which can lead to seizures, severe skin rashes and neurological problems, can be prevented by over-the-counter vitamins.

"We don't want to wait until they arise to treat them," he added.

Scott said that as well as pinpointing conditions, the programme, which is due to begin next year, will give an idea of public attitudes towards the lifetime storage of genomic data.

Potentially, the information could be used to help with an individual's future healthcare needs "to predict to diagnose or to treat conditions, for example, if they fall sick later on", he added.

—Inequalities—

The researchers intend to recruit from a broad spectrum of participants from different backgrounds, and in whom there is not necessarily an identified pre-existing risk.

"That crucially means that many of the parents we're approaching won't have any prior knowledge necessarily of genetics or inherited conditions in their family," said Amanda Pichini, a genetics counsellor at Genomics England.

If the trials prove successful, researchers aim to sequence the entire genome of newborns, alongside the existing heel-prick tests already carried out for nine rare but serious diseases in newborns, including sickle cell disease and cystic fibrosis.

UK Health Secretary Steve Barclay said "the potential for genomics to revolutionise the way we deliver health care is great".

"If we can detect treatable illnesses earlier and ensure patients access potentially lifesaving treatment faster, we could improve people's lives across the country, including thousands of babies, through this new pilot," he added in a statement.

His department highlighted the results of a public consultation, published in July 2021, indicating support for the use of genome sequencing in newborns if strong safeguards are in place.

Two other research programmes were also announced: one, with £22 million of government funding, is to sequence the genomes of up to

25,000 participants from non-European backgrounds, who are "currently under-represented" in research.

It aims to better understand DNA and its "impact on health outcomes" and to help "reduce health inequalities and level up patient outcomes across all communities", according to the Department of Health and Social Care.

The other, with initial funding of £26 million, focuses on the evaluation of sequencing to improve accuracy and speed in cancer diagnosis.

In total the government has announced £175 million in funding for genomics research.